

(19) World Intellectual Property
Organization
International Bureau



(43) International Publication Date
13 October 2005 (13.10.2005)

PCT

(10) International Publication Number
WO 2005/095955 A1

- (51) International Patent Classification⁷: **G01N 33/50**, 33/92 (74) Agent: A.P.T. Patent and Trade Mark Attorneys; PO Box 222, Mitcham, S.A. 5062 (AU).
- (21) International Application Number: PCT/AU2005/000461 (81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SM, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (22) International Filing Date: 31 March 2005 (31.03.2005)
- (25) Filing Language: English
- (26) Publication Language: English
- (30) Priority Data: 2004901726 31 March 2004 (31.03.2004) AU (84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IS, IT, LT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).
- (71) Applicant (for all designated States except US): **CHILDREN, YOUTH AND WOMEN'S HEALTH SERVICE** [AU/AU]; 72 King William Road, North Adelaide, S.A. 5006 (AU).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): **MEIKLE, Peter** [AU/AU]; 31 Minnamurra Drive, Redwood Park, S.A. 5067 (AU). **HOPWOOD, John** [AU/AU]; 2 Monarto Court, Stonyfell, S.A. 5066 (AU). **FULLER, Maria** [AU/AU]; 4/6 Redmond Street, Collinswood, S.A. 5081 (AU). **WHITFIELD, Phillip** [—/GB]; Faculty of Veterinary Science, University of Liverpool, Crown Street, Liverpool L69 7ZJ (GB). **SHARP, Peter** [AU/AU]; 18 Palmer Avenue, Myrtle Bank, S.A. 5072 (AU).
- Declaration under Rule 4.17:**
— of inventorship (Rule 4.17(iv)) for US only
- Published:**
— with international search report
- For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

(54) Title: SCREENING FOR LYSOSOMAL STORAGE DISEASE STATUS

(57) Abstract: A method of ascertaining the LSD (Lysosomal storage disorder) status of an individual. The method comprises taking a tissue or body fluid sample from the individual, estimating a level in the sample of each of three or more compound indicators. The indicators reflect the level of respectively each of three or more lipid containing storage associated compounds. The levels are used to calculate an LSD index number which is then compared with a standard to provide an assessment of the LSD status of the individual. The indicator compounds are conveniently phospholipids, glycolipids or lipopolysaccharide species measured by mass spectrometry. The method may be used to ascertain the nature of the disorder that the individual suffers from, and its severity. It may be used to monitor the progress of treatment and to ascertain the prospects of an individual contracting an LSD by providing a subclinical indicators for the condition.



WO 2005/095955 A1